ORIGINAL ARTICLE

PEDIATRIC POSTERIOR MIDLINE LUMPS: CNS MALFORMATIONS OR MASQUERADING DISEASE - MRI INSIGHTS

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PJR January - March 2024; 34(1): 11-17

ABSTRACT

INTRODUCTION: Central nervous system malformationsare frequently encountered anomalies owing to derangements in the neurulation process of spinal in the embryonic period. These are broadly categorized into 1) open spinal dysraphism and 2) closed spinal dysraphism, based on skin/sub cutaneous tissues coverage. Masquerading diseases may present with similar clinical presentation. Neuro-imaging plays an essential role in differentiating between neural tube defects and mimicking disease hence facilitate appropriate diagnosis and pre-surgical planning. **OBJECTIVE:** To assess the frequency of congenital central nervous system (CNS) malformations and masquerading diseases in the pediatric patients presenting with congenital posterior midline lumps at the head, neck and back using MRI brain and spine in the Radiology department of the national institute of child health (NICH). **METHODS: Design:** This is a descriptive, retrospective cross-sectional analysis. **Setting:** Radiology department of national institute of child health (NICH). Electronic data from November 2021 to October 2022 of MRI brain and spine of pediatric patients below 15 years presented with congenital posterior midline lumps were collected and analyzed on SPSS version 26. **RESULTS:** Total 201 pediatric patients were evaluated. There was a balanced gender distribution between male 91(45%) and female 110(54.7%). The most common presenting complaint was congenital lumbar posterior midline lump. Myelomeningocele was the most common diagnosis (61.2%) followed by lipomyelocele and lipomyelomeningocoele (4%). Notably, the most common association was found to be hydrocephalus (38.8%), syrinx (23.9%) and tethered cord (20.9%). Most prevalent masquerading disease was sacrococcygeal teratoma then macrocystic lymphatic malformation. Soft tissue hemangioma, hematoma and neurogenic tumor were also found in few patients. **CONCLUSION:** This study emphasizes the significance of neuroimaging detection of not only spinal dysraphism but other masquerading diseases as well. Early recognition of key imaging features of such malformations and mimicking diseases facilitates appropriate intervention hence plays acrucial role for therapeutic strategy and clinical outcome.
Keywords: Frequency, CNS malformations, neuroimaging and spinal dysraphism.

Introduction

Central nervous system malformations are frequently encountered anomalies with 1/100 live birth incidence. Spinal dysraphism is a collective term for congenital malformation of spine and spinal cord. It consists of diverse range of abnormality due to improper closure of midline structures. This includes anterior, posterior

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Submitted 2 January 2024, Accepted 24 January 2024

spinal dysraphism, dysraphism confined to the spinal canal and multi-compartment involvement and non-dysraphic masses of spinal region.

It is very difficult to make a diagnosis only on clinical based findings. In neonates and infants with suspected spinal and paraspinal anomalies, magnetic resonance imaging (MRI) remains the imaging gold standard and it is of immense importance in making diagnosis.[1]

Spinal dysraphism involves a spectrum of congenital anomalies resulting in a defective posterior elements through which meninges or neural elements are herniated, leading to a variety of clinical manifestations. According to clinical-radiologic classification, they are classified into 1) open spinal dysraphism (SDs) and 2) closed spinal dysraphism (SDs). Open spinal dysraphism (SDs) are usually associated with neural tissues and meningeal exposure to the external environment. Hence, there is an impending risk of CSF. Whereas the closed SDs have normal overlying skin, therefore, there is no neural placode exposure to the external environment.[2]

Closed spinal dysraphism is further divided into two groups: 1) closed SDs having subcutaneous mass and 2) closed SDs without subcutaneous mass.

Closed SDs having subcutaneous mass includes lipomyelocele, lipomyelomeningocoele, myelocystocele, meningocele. Closed SDs that lacks a subcutaneous mass is further divided into simple dysraphic state like filar / intradural lipoma, tight filum terminale and persistence of terminal ventricle and complex dysraphic states like dorsal dermal sinus, diastometamyelia, caudal regression syndrome and limited dorsal myeloschisis.[20]

The degree of neural placode malformation and level of the posterior element defect depicts the extent and severity of neurology deficit. The higher the level, usually worse is the prognosis. The most affected spinal region is lumbar region. A spectrum of neurological abnormalities like hydrocephalus, diastometamyelia, Arnold Chiari malformation, syrinx, gyral malformations, uro-vesical defects and skeletal abnormalities can be associated.[3] Multiple other associations can also be seen like spinal lipoma, tethered cord, myelocystocele, spinal dermoid / epidermoid, neuroenteric cyst, vertebral duplication, dorsal dermal sinus, split cord, Dandy Walker, Arnold Chiari, hydrocephalus, corpus callosum agenesis, syrinx, caudal regression, subcutaneous lipoma, neurogenic bladder etc. MRI is still the most preferable imaging tool particularly in those patients for spinal dysraphism.[8]

Masquerading diseases such as macro cystic-lymphatic malformation, cervical teratoma and sacrococcygeal mass like sacral chordoma or rhadomyosarcoma may present with similar presenting complaint of congenital posterior midline lump or poor urinary control.[23-24] The objective of this study is to evaluate the frequency of central nervous system (CNS) malformations, associations and other masquerading disease in pediatric patients presenting with congenital posterior midline lump using brain and spine MRI. This will not only signify the pivotal role of neuroimaging for early recognition of CNS malformation and mimicking diseases but also facilitates timely appropriate intervention, thereby resulting in effectiveness of the therapeutic strategy and the surgical planning.

Materials and Methods

It is a descriptive, retrospective cross-section analysis. Electronic data of MRI brain and spine of pediatric patients presenting with congenital posterior midline lumps at the head, neck and back in Radiology department of national institute of child health (NICH), from November 2021 to October 2022 were evaluated. Imaging was conducted on MRI machine Canon 1.5 Tesla, software version 6.1. Total 201 pediatric patient of age ranging between births to 15 years presented with congenital posterior midline lumps were included. Post-surgical cases were excluded from study.

Data were analyzed on SPSS 26. Quantitative analysis of gender and frequencies of each cerebral, spinal malformation, their associations and masquerading diseases were recordedand presented in tabulated and graphical format.

Results

The study included 201 pediatric patients for a period of one year retrospectively below 15 years. Out of total 201 patients, age groupings were as follows 137 (68.2%) belonged to 0-6 months-age group. 19 (9.5%)

instances were between 7-12 months age group, 30 (14.9%) cases were between 1-5years, 10 (5%) were between 6-10 years and 5 (2.5%) belonged to 11-15 years age group. (Tab.1)

Age Group	Frequency	Percent
0 - 6 Months	137	68.2
7 - 12 Months	19	9.5
1 - 5 Years	30	14.9
6 - 10 Years	10	5.0
11 - 15 Years	5	2.5
Total	**201**	**100.0**

Table 1:

Among these patients 91 (45%) were male and 110 (54.7%) were female which displayed approximately a balanced gender distribution. (Tab.2)

Gender	Frequency	Percent (%)
Male	91	45.3
Female	110	54.7
Total	**201**	**100.0**

Table 2:

The most common presenting complaint was lumbar posterior midline lump (74.6) followed by cervical (7%), sacral (7%), occipital posterior midline lump (4%,). The other less frequent presenting complaints were increasing head size (1.5%), poor urinary control (1.5%). Increase head size and posterior midline lump also presented simultaneously (4%). Very few patients were presented with dorsal midline lump and melanocytic nevus (1%). (Tab.2)

In the result of MRI, open spinal dysraphism account for 72% followed by close spinal dysraphism (14%) and mimicking disease (14%). Myelomeningocele was the most common diagnosis (61.2%) followed by lipomyelocele (4%) and lipomyelomeningocoele (4%). For these malfromations, lumbar region was the most affected location followed by cervical region. Complex dysraphic states including dorsal dermal sinus, diastometamyelia and caudal regression syndrome account for 3.5%. Intra dural lipoma or simple dysrahic state without subcutaneous mass was found in only 0.5%.

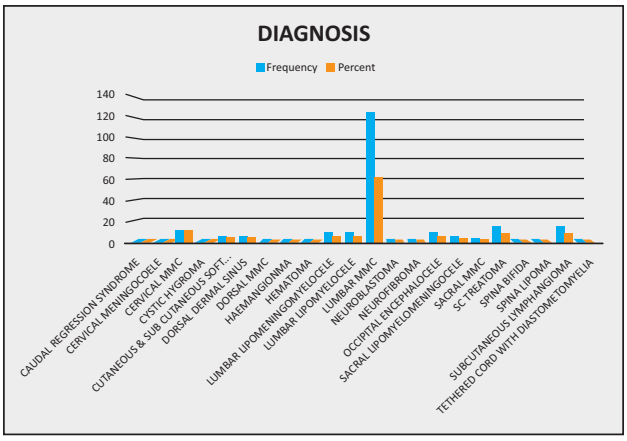

Table 3:

Most common masquerading disease was sacro-coccygeal teratoma presented with midline sacro-coccygeal mass (7%) followed by macrocystic lymphatic malformation (2.5%) and non specific cutaneous subcutaneous soft tissue thickening (2%). Soft tissue hemangioma (0.5%), hematoma (0.5%), neuroblastoma (0.5%) and neurofibroma (0.5%) was also found in some patients.

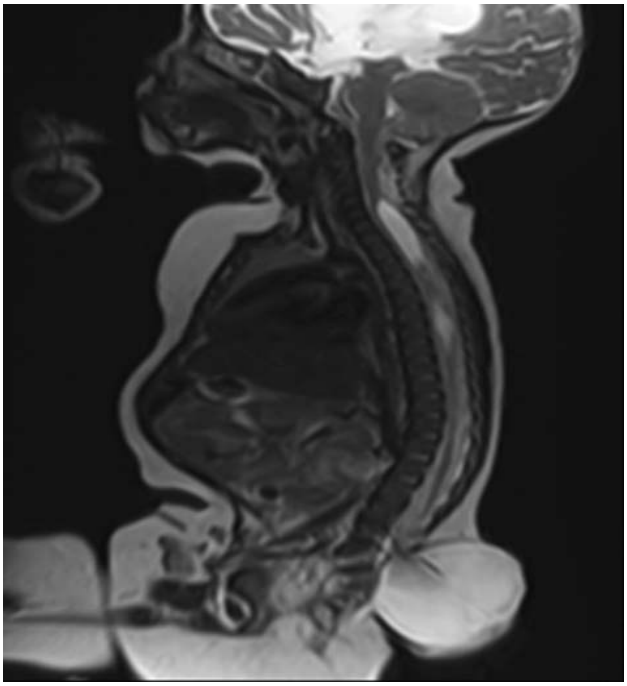

Figure 1: MRI whole spine,T2WI, sagittal section demonstrate defective neural arch of lower lumbar vertebrae with herniation of CSF filled sac along with neural placode without skin and subcutaneous tissues covering correspond to lumbar myelomeningocele. Note the associated syrinx in the upper dorsal cord.

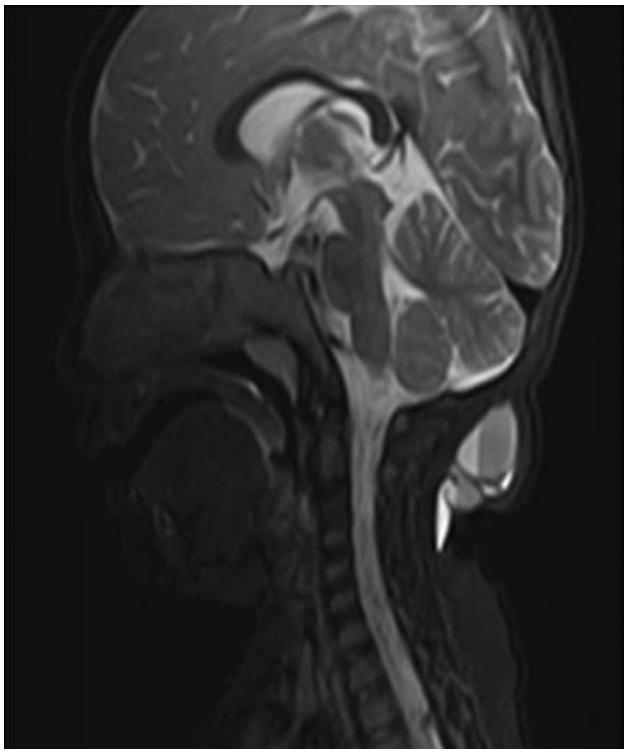

Figure 2: MRI cervical spine, T2WI, sagittal section demonstrates a well delineated subcutaneous cystic lesion at the nape of neck in midline with fluid-fluid levels without evidence of direct communication with spinal canal and any surrounding aggressive changes inferring macro-cystic lymphatic malformation.

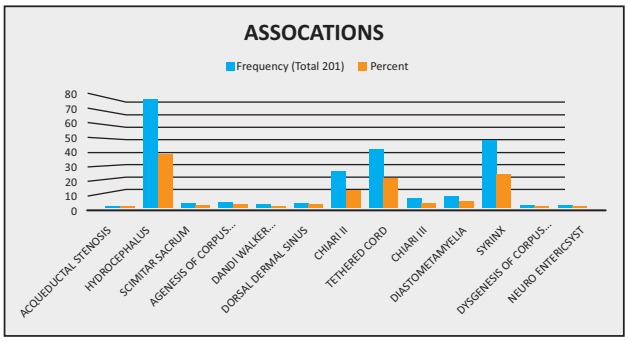

Table 4:

Notably, the most common association was found to be hydrocephalus (38.8%) followed by syrinx (23.9%) and tethered cord (20.9%). Other less prevalent association Chiari II (12.9%), Chiari III (3.5%) and diastometamyelia (4%) were also noted. Less frequent association like agenesis/dysgenesis (3%), scimitar sacrum (1.5%), Dandy Walker Malformation (0.5), Aqueduct Stenosis (0.5) and neuro enteric cyst (0.5) were noted in decreasing order of frequency.

Discussion

The incidence of spinal malformation has been decreasing for several decades. This is primarily due to appropriate antenatal care, adequate supplementary intake of folic acid with maternal nutrition, antenatal ultrasonography and genetic screening.[4,5,6,20] The incidence of most common, myelomeningocele (MMC) also varies worldwide; for example, the incidence among continental European countries varies between 1 in 1700 to 10,000 live births, whereas the incidence in the British Isles varies from 1 in 260 to 400. Ireland has the highest rate in Europe, at 1 in 200 live births. Neural tube defects is not uncommon in our local population. Its clinical manifestations are in line with international reported literature. The most acceptable reason is that our population face difficulty to access health care facility. Many pregnant women undergoes unsupervised pregnancy and are least aware of adverse effects of these congenital malformations outcome.[21]

In our study, the most frequent clinical presentation was congenital posterior midline lump at lumbar region followed by cervical/sacral region. The other less frequent complaint were simultaneous increase head size with lumbar posterior midline lump (2.5%). In few cases delayed mile stones (2.5%) and congenital melanocytic nevus with focal posterior midline lump (0.5%) at lumbar region was noted.

Ruangtrakool et al.documented incontinence of feces and urine, gait abnormality too.[20] In our study, poor urinary control was found to be in 1.5% of patients. Multiple cutaneous stigmata like presence of abnormal tuft of hair, sinus opening, subcutaneous lumps and cutaneous pigmentations also give awareness about such underlying neural tube congenital malformations. There is an analogous relation with VACTERL spectrum and tethered cord syndrome as well.[7] This study showed cutaneous manifestation of abnormal hair tuft and sinus opening in 6.5% of patients. Usually, these malformations are diagnosed in perinatal period but mostly closed spinal dysraphism are diagnosed late, due to absence of cutaneous stigmata. Hence, MRI becomes necessary for evaluation of these dysraphic state due to its better soft tissue characterization and multi-parametric imaging capabilities.[18-19]

In this study, aforementioned results showing involvement of spinal segment are comparable with the results of Tawfik et al.[12] and Kommana et al.[10] These showed that lumbar spine the most common affected region for spinal malformation 71.1% and 28.5%. Their study showed the lumbar region was followed by dorsolumbar spine. Whereas Dhigani et al demonstrated that lumbar region is the most commonly affected region (52.6%) followed by sacrococcygeal spine (34.2%).[13] In our study, lumbar spinal dysraphism was most affected region followed by cervical and then sacrococcygeal spine. Spinal dysraphism vary in appearance, size, and location; neurologic impairment generally is related to a larger size and more cranial location.

Blom HJ et al showed that other associated malformations are also common and include Arnald Chiari malformation in 98%, hydrocephalus in 70%, syrinx in 80% and low-lying spinal cord in almost all patients.[9,10] Our study demonstrates the notable common association hydrocephalus (38.8%) followed by syrinx (23.9%) and tethered cord (20.9%). Contrary to the Blom HJ study, Chiari II malformation was found in (12.9%). In decreasing order of frequency, other associations of Chiari II malformation (12.9%), Chiari III malformation (3.5 agenesis/dysgenesis (3%), scimitar sacrum (1.5%), Dandy Walker malformation (0.5), aqueduct stenosis (0.5) and neuro enteric cyst (0.5) were also recorded.

Both Tawfik et al.[12] and Dhingani et al.[13] showed comparable similar prevalence of the aforementioned CSD subtypes, apart from the mere 6.6% for tethered cord in Tawfik et al. as opposed to the compelling 60% in our own cohort. Whereas in our study, tethered cord was found to be 20.9%.

Non-spinal associated anomalies confronted in our study varied in diversity and frequentness compared to other studies as discussed in detail in results section.[14-20] Like in our study most common masquerading diseases that presented with congenital posterior midline mass was sacrococcygeal teratoma (7%) followed by macrocystic lymphatic malformation (2.5%) and nonspecific cutaneous subcutaneous soft tissue thickening (2%). Some other diseases with similar presenting complaints were soft tissue hemangioma (0.5%), hematoma (0.5%), neuroblastoma (0.5%) and neurofibroma (0.5%).

Despite scarcity in the literature regarding morphological appearance of lumbosacral and coccygeal regions, diverse variation in the clinical presentation forms a diagnostic conundrum while screening neonates. Hence, the role of neuroimaging in such instances becomes essential.[15]

Conventional radiographs are screening modality for bony spinal anomalies. In antenatal period, sonography is a screening modality, but it is less sensitive for closed spinal dysraphism. Computed tomography (CT) with multiplanar reformatted images is excellent for bony details like bony septum in Diastematomyelia and spinal anomalies. But CT provides less soft tissue details and requires instillation of non-ionic, iodinated contrast into thecal sacfor proper evaluation of theca and its contents. CT as well as Ultrasound don t have tissue contrast as good as MRI, hence, MRI is modality of choice for detailed evaluation of spinal dysraphism and masquerading disease. For MRI proper protocol is essential. It is time-consuming and inadequately available. There is also requirement of sedation in many children. Despite these constraints, MRI is the most preferred modality for assessment of spinal dysraphism and similar diseases of spine due to its excellent soft tissue contrast evaluation.[16-17]

Trap B et al mentioned in his study that congenital malformations of spinal cord and similar diseases differ significantly in clinical and imaging features and therefore, it is a diagnostic conundrum even for skilled radiologists. MRI makes it possible due to its excellent multiplaner capability and soft tissue contrast resolution thereby allowing radiologist to evaluate the fine details of such malformations. Accurate early detection of such conditions is of utmost importance related to good clinical outcome, which allows not only allows counselling of parents, taking appropriate therapeutic decision but also reduces morbidity related to congenital malformations.[5,20]

Limitation of this study is that it s a single institution-based study and doesn t have any base line study of similar type. With a larger sample size, such study should be conducted not only for the assessment of prevalence of these malformations and masquerading diseases but also should be addressed at multi-levels for better clinical outcome.

Conclusion

This study highlights that spinal dysraphism is not

uncommon in our local population and underscores the utmost importance of neuroimaging to distinguish between common entity spinal dysraphism and mimicking diseases like sacrococcygeal teratoma, macro cystic lymphatic malformation, hemangiomas, neuroblastoma and neurofibroma. Hence, knowledge of the important imaging findings is essential for the radiologist to effectively address spinal dysraphism and masquerading diseases for optimizing therapeutic strategy, surgical planning and better clinical outcome.

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